Cerebrofacial Venous Anomalies, Sinus Pericranii, Ocular Abnormalities and Developmental Delay

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Summary

The clinical implications of venous cerebrovascular maldevelopment remain poorly understood. We report on the association of cerebrofacial venous anomalies (including sinus pericranii), ocular abnormalities and mild developmental delay in two children. In addition, one child had a seizure disorder. Complex cerebrofacial slow-flow vascular anomalies may herald an underlying developmental aberration affecting the cerebrofacial and orbital regions.

Introduction

The coexistence of intracranial and extracranial vascular anomalies has been well documented in the literature [1-3]. Abnormal development of the brain has also been associated with anomalies of the cerebral and facial vessels [4-7]. Sinus pericranii refers to an abnormal communication between the intracranial dural venous sinuses and anomalous epicranial veins. In this communication, we describe two patients with cerebrofacial venous anomalies including sinus pericranii, in association with ocular abnormalities, mild developmental delay and possibly seizures.

Case Reports

Institutional review board approval was not required by our hospital for publishing retrospective case reports. However, in keeping with the ethical conduct of studies, the principles of the Declaration of Helsinki were followed.

First Patient

An eight-year-old boy was referred to our center for treatment of cerebrofacial vascular anomalies. The patient was born with a vascular "birthmark" on the forehead and around the left eye, and he had a medically-controlled seizure disorder. Examination showed a small left orbit and globe, with compromised visual acuity, proptosis, ptosis, glaucoma, miosis and dense retropupillary membranes (Figure 1A). There was a palpable bony defect in the forehead with overlying compressible, non-pulsatile bluish soft tissue which distended in the dependent position. The child had academic difficulties in reading and writing, with attention deficit disorder.

Cross-sectional studies demonstrated a smaller left cerebral hemisphere, with focal calcification and a cavernous malformation in the basal ganglia (Figure 1B,C). The venous malformation in the left orbit and frontal region

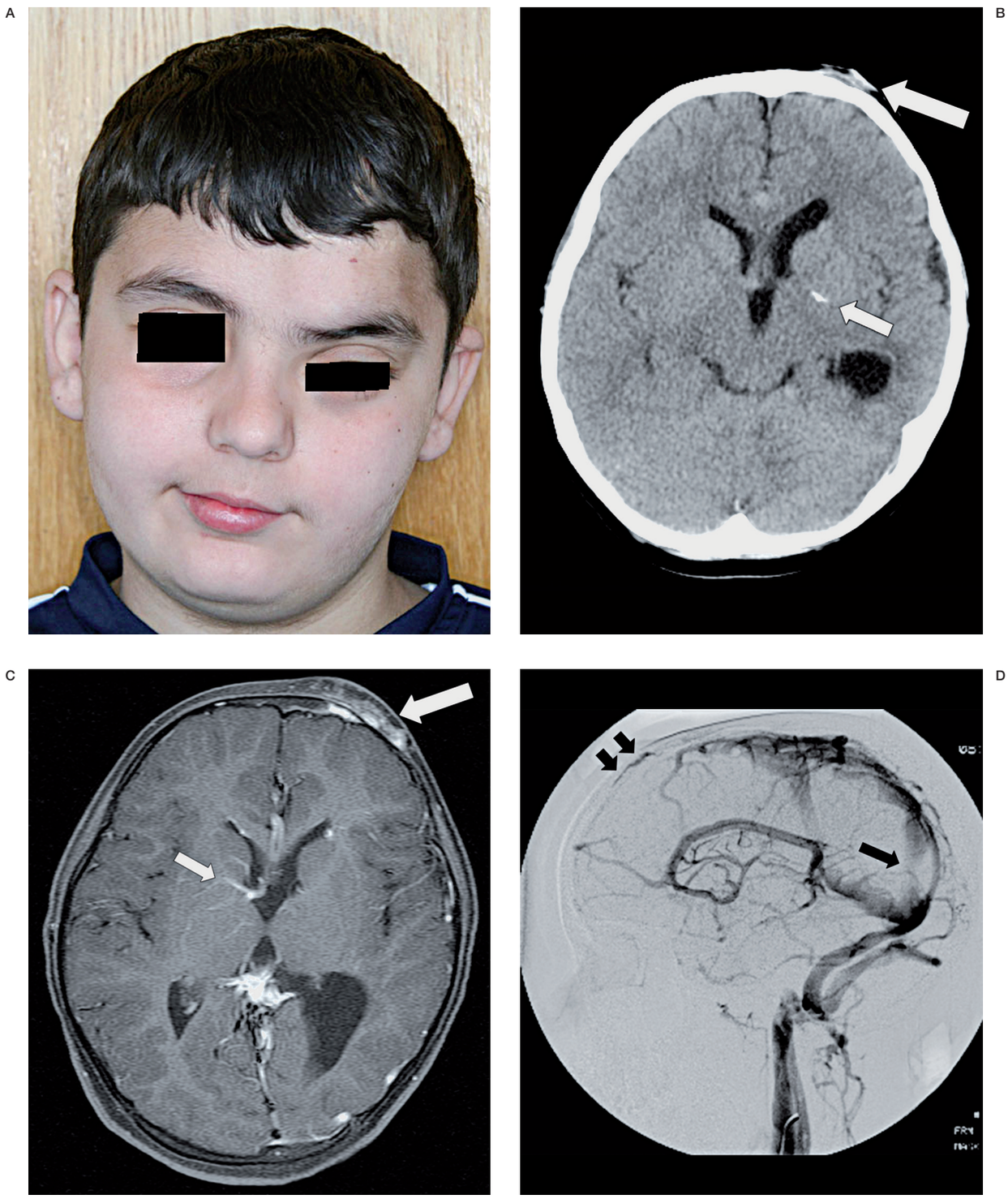
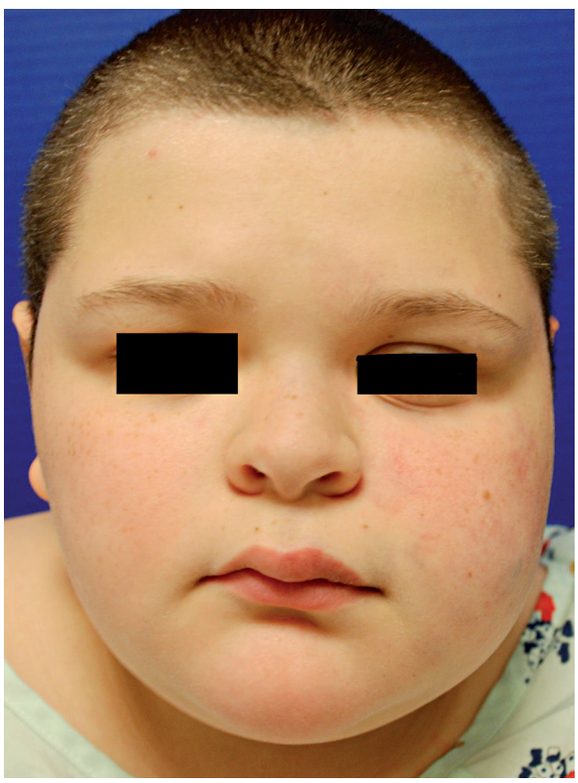
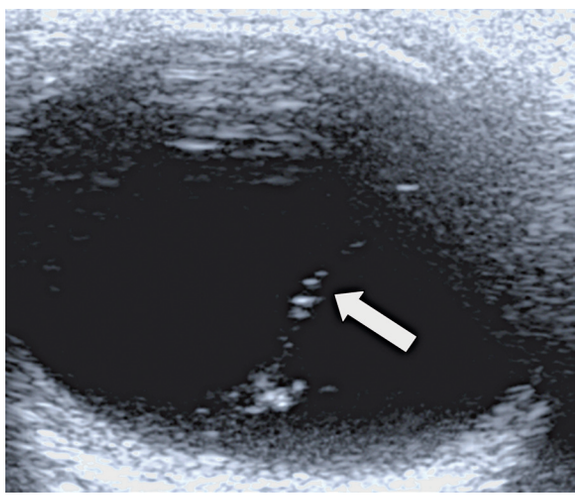


Figure 1 A) First Patient. There is a small left orbit and globe with venous expansion of the left forehead. B) Non-contrast enhanced head CT scan reveals a focal calcification in the left basal ganglia (small arrow) and venous malformation of the scalp (large arrow). C) Enhanced Axial T1-weighted MR Image shows smaller left cerebral hemisphere with DVAs in the right caudate nucleus and thalamus (small arrow). The anomalous transcalvarial and epicranial veins are clearly depicted (large arrow). D) Venous phase of cerebral angiography shows global anomalies of the venous drainage of the brain. Note the presence of persistent falcine sinus (long arrow) and fenestrated, underdeveloped superior sagittal sinus with retrograde filling of the sinus pericranii (short arrows).





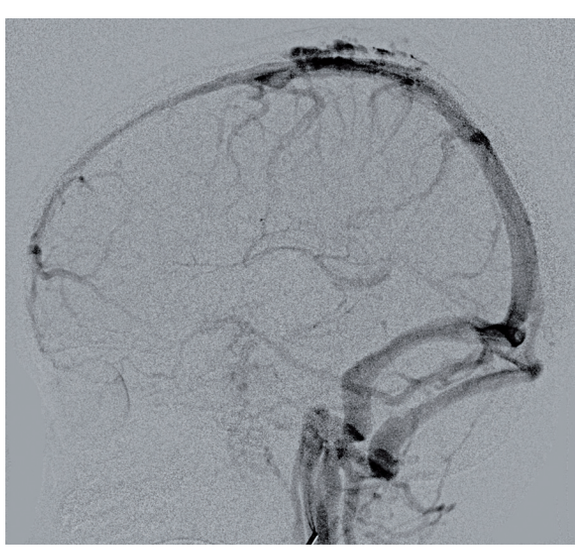


Figure 2 A) Second Patient. An overgrown left cheek with venous malformation extending into the upper lip and upper eyelid. B) Sonographic image of the left orbit demonstrates linear echogenic septa (arrow) within the posterior aspect of the vitreous space. C) Venous phase of cerebral angiography show transcalvarial communication between the superior sagittal sinus and the epicranial venous malformation.

traversing the left frontal calvaria represents a sinus pericranii. There were multiple complex developmental venous anomalies (DVA) in the left cerebral hemisphere. Cerebral angiography demonstrated several anomalies of the venous drainage of both cerebral hemispheres, including the presence of persistent falcine sinus and fenestrated, underdeveloped superior sagittal sinus with retrograde filling of the sinus pericranii.

Second Patient

A three-year-old boy was born with a vascular "birthmark" on the left cheek that progressively enlarged. A small bony groove was felt on the left side of the calvaria at the age of one year. The child had minor developmental delay with attention deficit disorder, learning disabilities, and delayed speech. There was a reticular blue stain overlying an enlarged left cheek, upper eyelid and oral mucosa. The cheek was soft and compressible. He had an irregular groove on the left parietal calvaria with large overlying venous channels.

MRI study showed a venous malformation of the left parietal calvaria with sinus pericranii extending into the left upper eyelid and masseter muscle (Figure 2A) with a left cerebral DVA. An ultrasonographic examination of the left orbit revealed linear echogenic membranes within the vitreous space with no significant flow within these membranes (Figure 2B). The visual acuity of the left eye was decreased, with ptosis and no retinal detachment. Cerebral an-

giography demonstrated normal arterial and cerebral venous anatomy with opacification of the sinus pericranii connecting the superior sagittal sinus through a parietal calvarial defect to anomalous tortuous scalp veins (Figure 2C).

Discussion

Sinus pericranii connects the extracranial and intracranial venous channels, typically in the midline frontal region [7-8]. Sinus pericranii is usually asymptomatic, although local pain, headache, nausea, and vertigo have been reported [9].

Sinus pericranii has been reported in association with variable slow-flow vascular anomalies of the cerebrofacial region, including cerebral and cerebellar cavernous malformation, cerebral DVAs, venous malformations of the tongue and blue rubber bleb nevus syndrome [7,10-12]. Konez et al. reported sinus pericranii in six out of 53 patients with cervicofacial venous malformation [13].

In addition to the cerebrofacial venous anomalies, both patients presented have mild developmental delay and one seizure disorder. Different forms of cerebrofacial vascular anomalies have been described in association with developmental delay including sinus pericranii, dural sinus malformation, cerebral "angiomas" and anomalous cerebral veins [14-16].

The coexistence of intracranial and extracranial vascular anomalies is not rare. Developmental venous anomalies are present in 20% of patients with extensive cervicofacial venous malformation [17]. Forty-five percent of patients with periorbital lymphatic malformation were found to have a cerebral DVA [2-3]. Bisdorf et al. [3] reported that 70% of orbital lymphatic malformation had intracranial vascular anomalies. Cerebrofacial venous metameric syndromes can be categorized into three main types: CVMS 1 (the medial prosencephalic or olfactory group with forehead and nose involvement), CVMS 2 (the lateral prosencephalic or optic group with involvement of the occipital lobe, eye, cheek, and maxilla) and CVMS 3 (the rhombencephalon or otic group with involvement of the cerebellum, lower face and mandible) [18-19]. Wyburn-Mason (or Bonnet-Dechaume-Blanc) syndrome is characterized by arteriovenous malformation of the brain, the orbit and the maxillofacial region [1]. The major features of Sturge-Weber syndrome are facial capillary stain, leptomeningeal and choroidal small vessel anomalies, mental retardation, seizure, weakness, visual impairment and glaucoma [20]. Bannayan–Riley–Ruvalcaba syndrome combines vascular anomalies (such as cerebral venous anomalies and fast-flow lesions) with variable degrees of developmental delay [21].

Vascular insults are implicated in abnormal cerebral development [4-6]. Barkovich [4] reported four patients with focal migrational anomalies associated with large vascular channels, three of them had developmental delay with seizure disorder. There are several case reports of the association of cortical migration disorder with arterial anomalies [22-25]. The association of developmental venous anomaly and neuromigrational disorders was reported by Riel-Romero et al. [25]. An early embryonic vascular aberration affecting the cervicofacial endothelium (mesodermal) or the tunica media (from the neural crest) may lead to migration anomalies with segmental distribution in the regions of the face and brain [1].

In conclusion, the combination of cerebrofacial vascular anomalies with sinus pericranii, ocular abnormalities, developmental delay and possibly seizure disorder might represent a rare association that has not been previously appreciated.

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